

Using NCBI's MedGen in Clinical Practice

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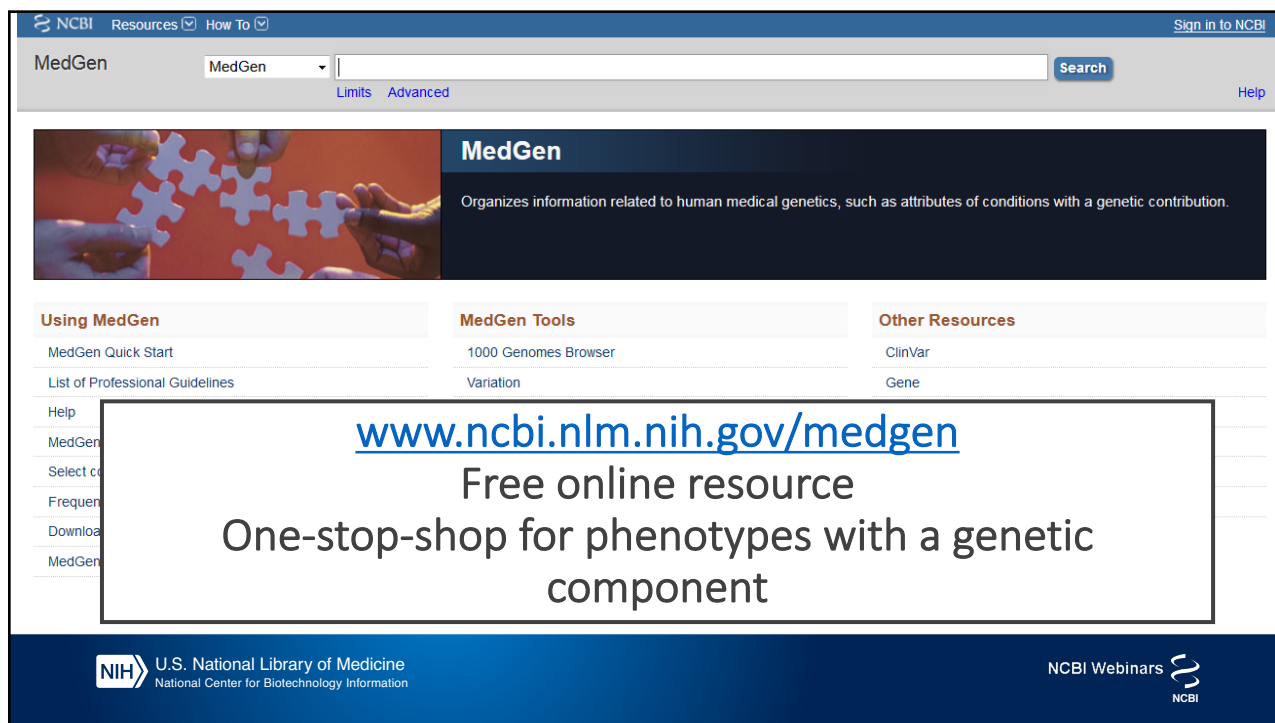
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Using NCBI's MedGen in Clinical Practice

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 Adriana.Malheiro@nih.gov

Goals

- Overview of MedGen
 - Understand its content
- Learn how to search and navigate MedGen
- Identify ways to incorporate MedGen in the clinical genetics process



NCBI Resources How To Sign in to NCBI


MedGen MedGen Search Limits Advanced Help

MedGen
Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.

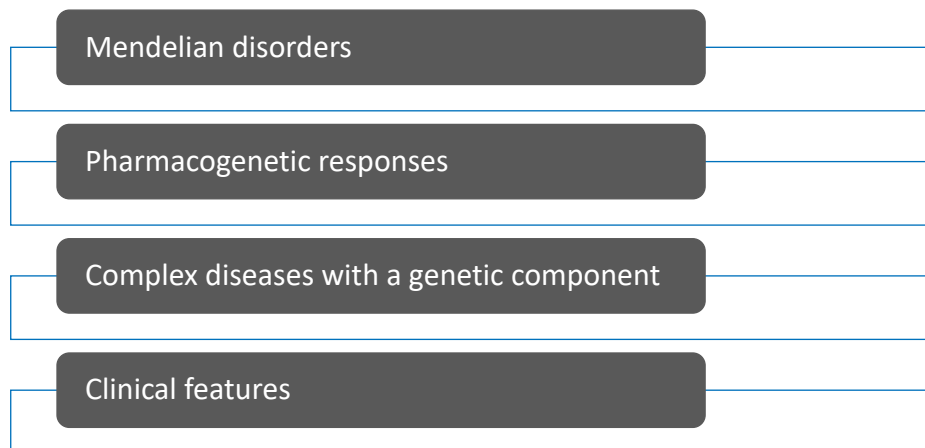
Using MedGen	MedGen Tools	Other Resources
MedGen Quick Start	1000 Genomes Browser	ClinVar
List of Professional Guidelines	Variation	Gene
Help		
MedGen		
Select c		
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Downloa		
MedGen		

www.ncbi.nlm.nih.gov/medgen
Free online resource
One-stop-shop for phenotypes with a genetic component

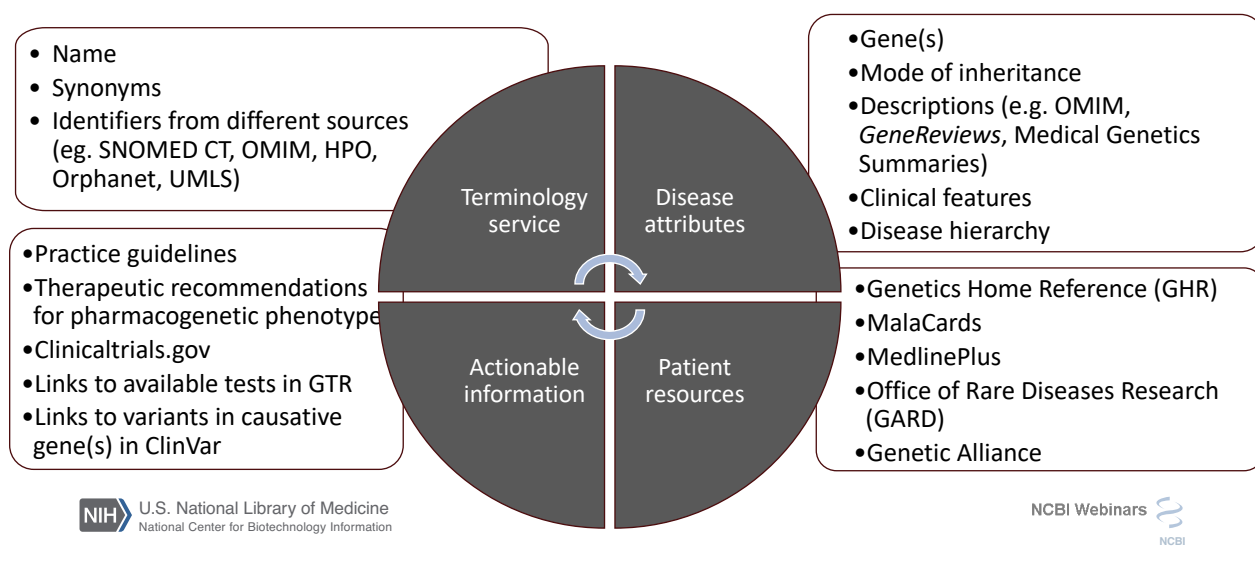
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NCBI Webinars 

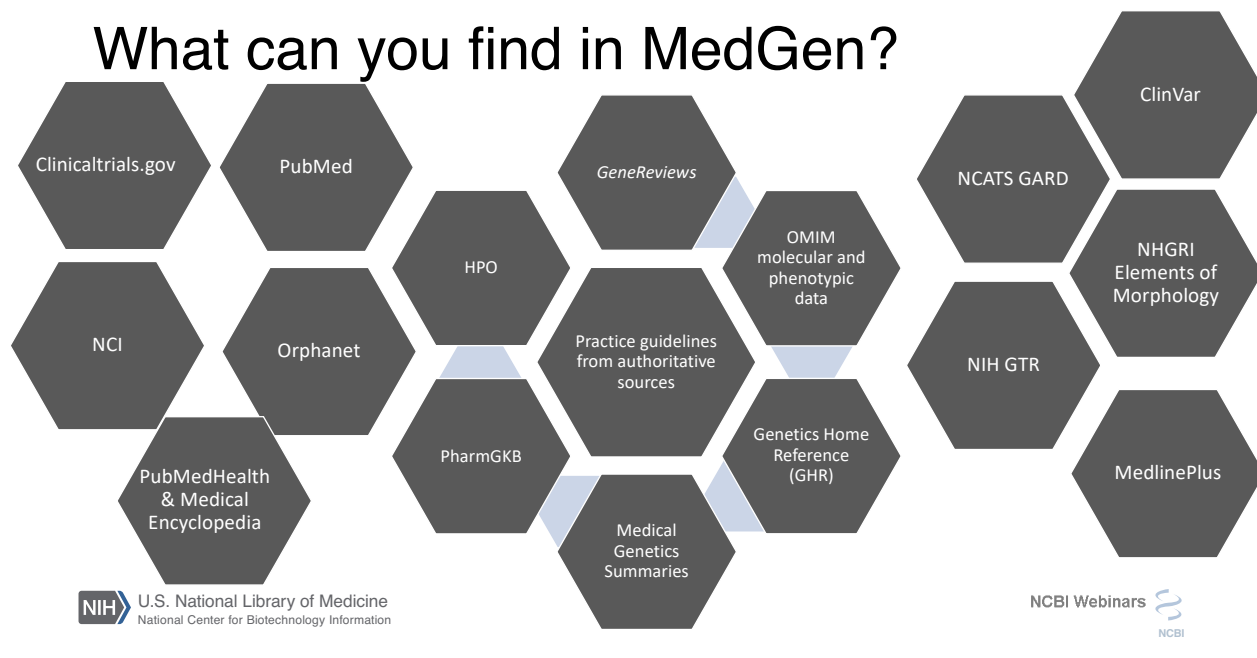
Types of records



What content is on a MedGen record?



What can you find in MedGen?



NCBI Resources How To Sign in to NCBI

MedGen MedGen Search Limits Advanced Help

MedGen
Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.

Using MedGen	MedGen Tools	Other Resources
MedGen Quick Start	1000 Genomes Browser	ClinVar
List of Professional Guidelines	Variation	Gene
Help		Genetic Testing Registry (GTR®)
MedGen Chapter in The NCBI Handbook		GeneReviews®
Select condition and phenotype terms for ClinVar and GTR		OMIM®
Frequently asked questions		RefSeqGene
Downloads/FTP		
MedGen News		

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Simple search

- Condition name & acronym
- Drug response
- Clinical feature
- Gene
- OMIM #

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MedGen

MedGen

Limits Advanced Help

MedGen

Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.

Using MedGen	MedGen Tools	Other Resources
MedGen Quick Start	1000 Genomes Browser	ClinVar
List of Professional Guidelines	Variation	Gene
Help		Genetic Testing Registry (GTR®)
MedGen Chapter in The NCBI Handbook		GeneReviews®
Select condition and phenotype terms for ClinVar and GTR		OMIM®
Frequently asked questions		RefSeqGene
Downloads/FTP		
MedGen News		

Example searches

Name	Related gene	Clinical feature
achondroplasia[title]	LMNB1[gene]	short stature[clinical features]

As you type your query, names of genetic disorders used in the NIH Genetic Testing Registry (GTR) will be provided. If you do not make a selection from the menu that appears under the search box as you type, your query is processed by looking for a match on a word or phrase. * is used as the wild card, and that wild card can be used only at the end of a word.

If you enter a gene symbol followed by [gene], the diseases caused by or with some association to that gene will be retrieved.

If you enter the name of the feature followed by [clinical feature] the diseases with that feature will be retrieved.

Search by clinical features

Get a list of conditions with the clinical feature derived from data from HPO and OMIM

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medline My NCBI Sign Out

MedGen MedGen [Arachnodactyly] Search

Create alert Limits Advanced Help

See MedGen results with Arachnodactyly as a clinical feature (55)

Summary 20 per page

Search results

Items: 1 to 20 of 70

Arachnodactyly

A tall and slim body build with increased arm span to height ratio (>1.05) and a reduced upper-to-lower segment ratio (<0.85), i.e., unusually long arms and legs. The extremities as well as the hands and feet are unusually slim. [from HPO]

MedGen UID: 2047 • Concept ID: C0003709 • Congenital Abnormality, Finding

GTR ClinVar Genes OMIM GeneReviews

1. Congenital contractural arachnodactyly

2. Congenital contractural arachnodactyly (CCA) is characterized by a Marfan-like appearance (tall, slender habitus in which arm span exceeds height) and long, slender fingers and toes (arachnodactyly). Most affected individuals have "crumpled" ears that present as a folded upper helix of the external ear and most have contractures of major joints (knees and ankles) at birth. The proximal interphalangeal joints also have flexion contractures (i.e., camptodactyly), as do the toes. Hip contractures, adducted thumbs, and club foot may occur. The majority of affected individuals have muscular hypoplasia. Contractures usually improve with time. Kyphosis/scoliosis is present in about half of all affected individuals. It begins as early as infancy, is progressive, and causes the greatest morbidity in CCA. Dilatation of the aorta is occasionally present. Infants have been observed with a severe/lethal form characterized by multiple cardiovascular and gastrointestinal anomalies in addition to the typical skeletal findings. [from GeneReviews]

MedGen UID: 67391 • Concept ID: C0200998 • Congenital Abnormality

GTR ClinVar Genes OMIM GeneReviews

3. Shprintzen-Goldberg syndrome

Shprintzen-Goldberg syndrome (SGS) is characterized by: craniosynostosis of the coronal, sagittal, or lambdoid sutures; dolichcephaly; distinctive craniofacial features; skeletal changes (dolichostenomelia, arachnodactyly; camptodactyly; pes planus; pectus excavatum or carinatum; scoliosis, joint hypermobility or contractures and C1/C2 spine malformation); neurologic abnormalities; intellectual disability; and brain anomalies (hydrocephalus; dilatation of the lateral ventricles; and Chiari I malformation). Cardiovascular anomalies may include mitral valve

Filter your results:

All (78)

Records in GTR (66)

Records in OMIM (65)

Diseases (75)

Records in Orphanet (33)

Records in HPO (1)

Recommended for clinicians (70)

Manage Filters

Find items

Search details

Arachnodactyly[All Fields]

Search

See more...

Recent activity

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NCBI

Clinical feature record

Arachnodactyly

MedGen UID: 2047 • Concept ID: C0003706 • Congenital Abnormality; Finding

Synonyms: Long slender fingers; Spider slender fingers

SNOMED CT: **Arachnodactyly** (62250003); Spider finger (62250003); Dolichostenomelia (62250003); Congenital arachnodactyly (62250003)

HPO: HP:0001166



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Definition

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Genetic Testing Registry

Deletion/duplication analysis (10)

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Definition

A tall and slim body build with increased arm span to height ratio (>1.05) and a reduced upper-to-lower segment ratio (<0.85), i.e., unusually long arms and legs. The extremities as well as the hands and feet are unusually slim. [from HPO]

Go to: (v) (v)

Conditions with the clinical feature: Arachnodactyly

Conditions with this feature

2p15-16.1 microdeletion syndrome
Achar syndrome
Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
Arterial tortuosity syndrome
Arthrogryposis, distal, with impaired proprioception and touch
Autosomal recessive cutis laxa type 1B
Camptodactyly, fibrous tissue hyperplasia, and skeletal dysplasia
Camptodactyly, tall stature, and hearing loss syndrome
Chromosome 10q22.3-q23.2 deletion syndrome
Chromosome 2q32-q33 deletion syndrome
Coffin-Siris syndrome 5
Congenital contractural arachnodactyly
Cutis laxa, autosomal recessive
Cutis laxa, autosomal recessive
Ehlers-Danlos syndrome progeroid type
Ehlers-Danlos syndrome, hydroxylysine-deficient
Ehlers-Danlos syndrome, musculocontractural type
Ehlers-Danlos syndrome, musculocontractural type 2
Epiphyseal chondrodysplasia, miura type
Frontometaphyseal dysplasia
Ham-Munk syndrome
Harrod-Doman-Kleele syndrome
Homocystinuria due to CBS deficiency
Leukodystrophy, hypomyelinating, 10

Marfan syndrome

MedGen UID: 44287 • Concept ID: C0024790 • Disease or Syndrome

Marfan syndrome, a systemic disorder of connective tissue with a high degree of clinical variability, comprises a broad phenotypic continuum ranging from mild (features of Marfan syndrome in one or a few systems) to severe and rapidly progressive neonatal multiorgan disease. Cardinal manifestations involve the ocular, skeletal, and cardiovascular systems. Ocular findings include myopia (the most common ocular feature), ectopia lentis (seen in approximately 60% of affected individuals), and an increased risk for retinal detachment, glaucoma, and early cataracts. Skeletal system manifestations include bone overgrowth and joint laxity, disproportionately long extremities for the size of the trunk (dolichostenomelia), overgrowth of the ribs that can push the sternum in (pectus excavatum) or out (pectus carinatum), and scoliosis that ranges from mild to severe and progressive. The major morbidity and early mortality in the Marfan syndrome relate to the cardiovascular system and include dilatation of the aorta at the level of the sinuses of Valsalva (predisposing to aortic tear and rupture), mitral valve prolapse with or without regurgitation, tricuspid valve prolapse, and enlargement of the proximal pulmonary artery. Severe and prolonged regurgitation of the mitral and/or aortic valve can predispose to left ventricular dysfunction and occasionally heart failure. With proper management, the life expectancy of someone with Marfan syndrome approximates that of the general population.

See: [Condition Record](#)

Myopathy, congenital, complex-north
Oculomeic amyoplasia
Osteopthia striata with cranial sclerosis
Progeroid facial appearance with hand anomalies
Pseudoaminoplerin syndrome
Radiohumeral fusions with other skeletal and craniofacial anomalies
Retinopathy pigmentary mental retardation
Severe X-linked myotubular myopathy
Shprintzen-Goldberg syndrome
Stickler syndrome type 1
Stickler syndrome, type 2
Willems-kolk syndrome
X-linked mental retardation with marfanoid habitus syndrome

Go to: (v) (v)

Reviews
PubMed Clinical Queries
Reviews in PubMed

Related information

ClinVar
GTR
GTR(Clinical)
MeSH

Recent activity

Arachnodactyly
Marfan syndrome
See more...

Advanced search: add multiple parameters

- Clinical features
- Genes
- Chromosomal locations
- Mode of inheritance
- OMIM#

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NCBI Resources How To Sign in to NCBI

MedGen MedGen Limits **Advanced** Search Help

NCBI Resources How To

MedGen Home Help

MedGen Advanced Search Builder

("arachnodactyly"[Clinical Features]) AND "aortic dissection"[Clinical Features]

Edit Clear

Builder

Clinical Features arachnodactyly[Clinical Features] Show index list

AND Clinical Features "aortic dissection"[Clinical Features] Hide index list

aortic dissection (5)
aortic regurgitation (23)
aortic root dilatation (6)
aortic tortuosity (1)
aortic valve atresia (1)
aortic valve calcification (4)
aortic valve stenosis (34)
aphakia (20)
aphakia, congenital primary (2)
aphalangy of hands and feet (1)

Previous 200
Next 200
Refresh index

AND Gene Name Show index list

Search or Add to history

MedGen MedGen ("arachnodactyly"[Clinical Features]) AND "aortic dissection"[Clinical Features] Search Create alert Limits Advanced Help

Summary

Search results

Items: 3

Marfan syndrome

Marfan syndrome, a systemic disorder of connective tissue with a high degree of clinical variability, comprises a broad phenotypic continuum ranging from mild (features of Marfan syndrome in one or a few systems) to severe and rapidly progressive neonatal multiorgan disease. Cardinal manifestations involve the ocular, skeletal, and cardiovascular systems. Ocular findings include myopia (the most common ocular feature); ectopia lentis (seen in approximately 60% of affected individuals); and an increased risk for retinal detachment, glaucoma, and early cataracts. Skeletal system manifestations include bone overgrowth and joint laxity; disproportionately long extremities for the size of the trunk (dolichostenomelia); overgrowth of the ribs that can push the sternum in (pectus excavatum) or out (pectus carinatum); and scoliosis that ranges from mild to severe and progressive. The major morbidity and early mortality in the Marfan syndrome relate to the cardiovascular system and include dilatation of the aorta at the level of the sinuses of Valsalva (predisposing to aortic tear and rupture), mitral valve prolapse with or without regurgitation, tricuspid valve prolapse, and enlargement of the proximal pulmonary artery. Severe and prolonged regurgitation of the mitral and/or aortic valve can predispose to left ventricular dysfunction and occasionally heart failure. With proper management, the life expectancy of someone with Marfan syndrome approximates that of the general population. [from GeneReviews]

MedGen UID: 44287 • Concept ID: C0024796 • Disease or Syndrome

GTR ClinVar Genes OMIM GeneReviews

1. **Loeys-Dietz syndrome 3**

2. Loeys-Dietz syndrome (LDS) is characterized by vascular findings (cerebral, thoracic, and abdominal arterial aneurysms and/or dissections), skeletal manifestations (pectus excavatum or pectus carinatum, scoliosis, joint laxity, arachnodactyly, talipes equinovarus, cervical spine malformation and/or instability), craniofacial features (widely spaced eyes, strabismus, bifid uvula / cleft palate, and craniosynostosis that can involve any sutures), and cutaneous findings (velvety and translucent skin, easy bruising, and dystrophic scars). Individuals with LDS are predisposed to widespread and aggressive arterial aneurysms and pregnancy-related complications including uterine rupture and death. Individuals with LDS can show a strong predisposition for allergic/inflammatory disease including asthma, eczema, and reactions to food or environmental allergens. There is also an increased incidence of gastrointestinal inflammation including eosinophilic esophagitis and gastritis or inflammatory bowel disease. Wide variation in the distribution and severity of clinical features can be seen in individuals with LDS, even among affected individuals within a family who have the same pathogenic variant. [from GeneReviews]

MedGen UID: 462437 • Concept ID: C3151087 • Disease or Syndrome

GTR ClinVar Genes OMIM GeneReviews

3. **Loeys-Dietz syndrome 4**

Filter your results:

All (3)
Records in GTR (3)
Records in OMIM (3)
Diseases (3)
Records in Orphanet (2)
Records in HPO (0)
Recommended for clinicians (3) Manage Filters

Find related data

Database: Select Find items

Search details

"arachnodactyly"[Clinical Features] AND "aortic dissection"[Clinical Features]

Search See more...

Recent activity

Turn Off Clear

Q ("arachnodactyly"[Clinical Features]) AND "aortic dissection"[Clinical Features] (3) MedGen

MedGen Disease Record

Full Report ▾

Send to: ▾

Marfan syndrome (MFS)

MedGen UID: 44287 • Concept ID: C0024796 • Disease or Syndrome

Synonyms: FBN1-Related Thoracic Aortic Aneurysms and Aortic Dissections; Marfan syndrome type 1; Marfan syndrome, classic; MARFAN SYNDROME, TYPE I; Marfan's syndrome; Marfanoid hypermobility syndrome; MFS

Modes of inheritance: Autosomal dominant inheritance (HPO, OMIM, Orphanet)

SNOMED CT: Marfan syndrome (19346006); Marfan's syndrome (19346006); Marfan's disease (19346006)

Gene (location): FBN1 (15q21.1)

OMIM®: 154700

Orphanet: ORPHA558

Disease characteristics

Go to: ☺ ☹ ☰

Excerpted from the GeneReview: Marfan Syndrome

Marfan syndrome, a systemic disorder of connective tissue with a high degree of clinical variability, comprises a broad phenotypic continuum ranging from mild (features of Marfan syndrome in one or a few systems) to severe and rapidly progressive neonatal multiorgan disease. Cardinal manifestations involve the ocular, skeletal, and cardiovascular systems. Ocular findings include myopia (the most common ocular feature), ectopia lentis (seen in approximately 60% of affected individuals), and an increased risk for retinal detachment, glaucoma, and early cataracts. Skeletal system manifestations include bone overgrowth and joint laxity; disproportionately long extremities for the size of the trunk (dolichostenomelia); overgrowth of the ribs that can push the sternum in (pectus excavatum) or out (pectus carinatum); and scoliosis that ranges from mild to severe and progressive. The major morbidity and early mortality in the Marfan syndrome relate to the cardiovascular system and include dilatation of the aorta at the level of the sinuses of Valsalva (predisposing to aortic tear and rupture), mitral valve prolapse with or without regurgitation, tricuspid valve prolapse, and enlargement of the proximal pulmonary artery. Severe and prolonged regurgitation of the mitral and/or aortic valve can predispose to left ventricular dysfunction and occasionally heart failure. With proper management, the life expectancy of someone with Marfan syndrome approximates that of the general population. [from GeneReviews]

Full text of GeneReview (by section):

Summary | Diagnosis | Clinical Characteristics | Genetically Related (Allelic) Disorders | Differential Diagnosis | Management | Genetic Counseling | Resources | Molecular Genetics | References | Chapter Notes

Authors:

Harry Dietz [view full author information](#)

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Mutation scanning of the entire coding region (6)

Sequence analysis of select exons (13)

Sequence analysis of the entire coding region (134)

Targeted variant analysis (23)

See all (170)

Clinical resources

Disease summaries from different sources (e.g. GeneReviews, OMIM, GHR)

Additional descriptions

Go to: ☺ ☹ ☰

From OMIM

A heritable disorder of fibrous connective tissue, Marfan syndrome shows striking pleiotropism and clinical variability. The cardinal features occur in 3 systems—skeletal, ocular, and cardiovascular (McKusick, 1972; Pyeritz and McKusick, 1979; Pyeritz, 1993). It shares overlapping features with congenital contractural arachnodactyly (121050), which is caused by mutation in the FBN2 gene (612570). Gray and Davies (1996) gave a general review. They published Kaplan-Meier survival curves for a cohort of British Marfan syndrome patients demonstrating greater survivorship in females than in males; a similar result had been reported by Murdoch et al. (1972) and by Silverman et al. (1995). Gray and Davies (1996) also proposed a grading scale for clinical comparison of the Marfan syndrome patients. The authors provided criteria for each grade and suggested uniform use of these scales may facilitate clinicomolecular correlations. <http://www.omim.org/entry/154700>

From GHR

Marfan syndrome is a disorder that affects the connective tissue in many parts of the body. Connective tissue provides strength and flexibility to structures such as bones, ligaments, muscles, blood vessels, and heart valves. The signs and symptoms of Marfan syndrome vary widely in severity, timing of onset, and rate of progression. The two primary features of Marfan syndrome are vision problems caused by a dislocated lens (ectopia lentis) in one or both eyes and defects in the large blood vessel that distributes blood from the heart to the rest of the body (the aorta). The aorta can weaken and stretch, which may lead to a bulge in the blood vessel wall (an aneurysm). Stretching of the aorta may cause the aortic valve to leak, which can lead to a sudden tearing of the layers in the aorta wall (aortic dissection). Aortic aneurysm and dissection can be life threatening. Many people with Marfan syndrome have additional heart problems including a leak in the valve that connects two of the four chambers of the heart (mitral valve prolapse) or the valve that regulates blood flow from the heart into the aorta (aortic valve regurgitation). Leaks in these valves can cause shortness of breath, fatigue, and an irregular heartbeat felt as skipped or extra beats (palpitations). Individuals with Marfan syndrome are usually tall and slender, have elongated fingers and toes (arachnodactyly), and have an arm span that exceeds their body height. Other common features include a long and narrow face, crowded teeth, an abnormal curvature of the spine (scoliosis or kyphosis), and either a sunken chest (pectus excavatum) or a protruding chest (pectus carinatum). Some individuals develop an abnormal accumulation of air in the chest cavity that can result in the collapse of a lung (spontaneous pneumothorax). A membrane called the dura, which surrounds the brain and spinal cord, can be abnormally enlarged (dural ectasia) in people with Marfan syndrome. Dural ectasia can cause pain in the back, abdomen, legs, or head. Most individuals with Marfan syndrome have some degree of nearsightedness (myopia). Clouding of the lens (cataract) may occur in mid-adulthood, and increased pressure within the eye (glaucoma) occurs more frequently in people with Marfan syndrome than in those without the condition. The features of Marfan syndrome can become apparent anytime between infancy and adulthood. Depending on the onset and severity of signs and symptoms, Marfan can be fatal early in life; however, the majority of affected individuals survive into mid- to late adulthood. <https://ghr.nlm.nih.gov/condition/marfan-syndrome>

Clinical resources

OMIM

Orphanet

ClinicalTrials.gov

Molecular resources

OMIM

View FBN1 variations in ClinVar

RefSeqGene

Coriell Institute for Medical Research

Consumer resources

Genetic Alliance

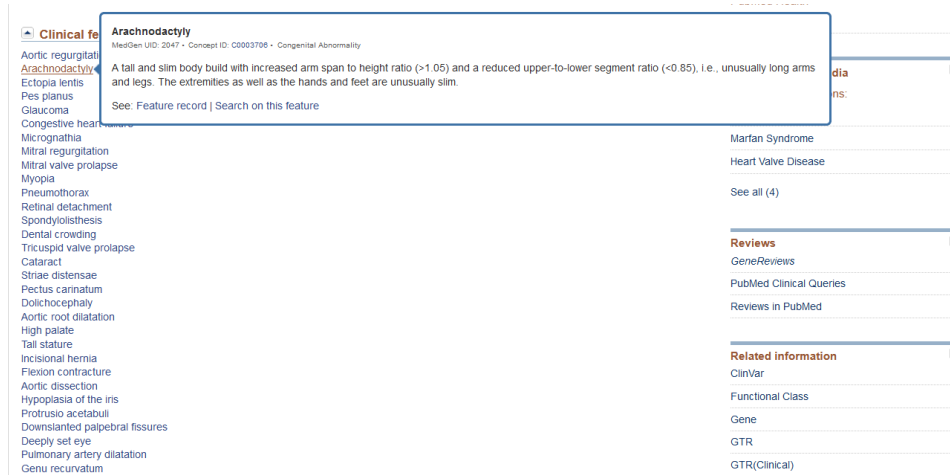
Genetics Home Reference

MalaCards

MedlinePlus

NCATS Office of Rare Diseases Research (GARD)

List of clinical features for the disease with descriptions (HPO, OMIM)



Clinical features

Arachnodactyly
MedGen UID: 2047 • Concept ID: C0003706 • Congenital Abnormality

A tall and slim body build with increased arm span to height ratio (>1.05) and a reduced upper-to-lower segment ratio (<0.85), i.e., unusually long arms and legs. The extremities as well as the hands and feet are unusually slim.

See: Feature record | Search on this feature

Related information

ClinVar

Functional Class

Gene

GTR

GTR(Clinical)

Reviews

GeneReviews

PubMed Clinical Queries

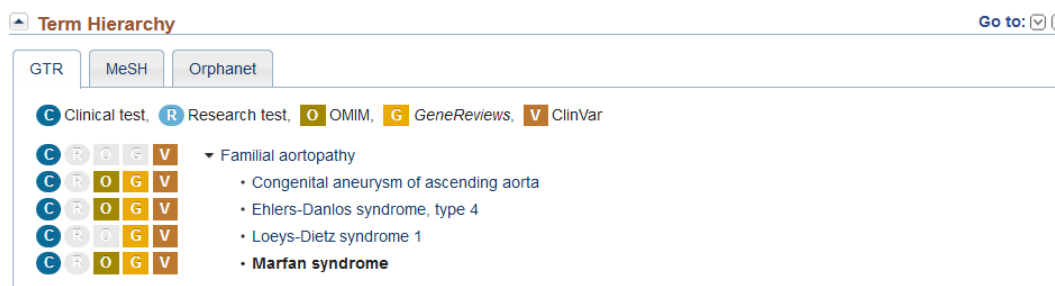
Reviews in PubMed

See all (4)

Marfan Syndrome

Heart Valve Disease

Disease hierarchy (manually curated, MeSH, Orphanet)



Term Hierarchy Go to: [] []

GTR MeSH Orphanet

C Clinical test, R Research test, O OMIM, G GeneReviews, V ClinVar

▼ Familial aortopathy

- Congenital aneurysm of ascending aorta
- Ehlers-Danlos syndrome, type 4
- Loeys-Dietz syndrome 1
- Marfan syndrome

Professional guidelines and suggested articles

Professional guidelines
Go to:

PubMed
[ACMG policy statement: updated recommendations regarding analysis and reporting of secondary findings in clinical genome-scale sequencing.](#)
ACMG Board of Directors.
Genet Med 2015 Jan;17(1):68-9. Epub 2014 Nov 13 doi: 10.1038/gim.2014.151. PMID: 25356965
[2014 ESC Guidelines on the diagnosis and treatment of aortic diseases: Document covering acute and chronic aortic diseases of the thoracic and abdominal aorta of the adult. The Task Force for the Diagnosis and Treatment of Aortic Diseases of the European Society of Cardiology \(ESC\).](#)
Erbel R, Aboyans V, Boileau C, Bossone E, Bartolomeo RD, Eggebrecht H, Evangelista A, Falk V, Frank H, Gaemperli O, Grabenwöger M, Haverich A, Iung B, Manolis AJ, Meijboom F, Nienaber CA, Roffi M, Rousseau H, Sechtem U, Simes PA, Almen RS, Vrints CJ; ESC Committee for Practice Guidelines.
Eur Heart J 2014 Nov 1;35(41):2873-926. Epub 2014 Aug 29 doi: 10.1093/eurheartj/ehu281. PMID: 25173340
[Canadian Cardiovascular Society position statement on the management of thoracic aortic disease.](#)
Boothwani M, Andelfinger G, Leipsic J, Lindsay T, McMurtry MS, Therrien J, Siu SC; Canadian Cardiovascular Society.
Can J Cardiol 2014 Jun;30(6):577-89. Epub 2014 Feb 28 doi: 10.1016/j.cjca.2014.02.018. PMID: 24882528
ACMG recommendations
Green RC, Berg JS, Grody W, et al.
Genet Med 2013 Jul;15(7):565-74. Epub 2013 Jun 11 doi: 10.1038/gim.2013.171. PMID: 23805451
Suggested Reading
Go to:

PubMed
[The revised Ghent nosology for the Marfan syndrome.](#)
Loeys BL, Dietz HC, Braverman AC, Callewaert BL, De Backer J, Devereux RB, Hilhorst-Hofstee Y, Jondeau G, Faivre L, Milewicz DM, Pyeritz RE, Sponseller PD, Wordsworth P, De Paepe AM
J Med Genet 2010 Jul;47(7):476-85. doi: 10.1136/jmg.2009.072785. PMID: 20591885
[Clinical utility gene card](#)
Arslan-Kirchner M, Arbustini E, Boileau C, Child A, Colod-Beroud G, De Paepe A, Epplen J, Jondeau G, Loeys B, Faivre L, et al.
Eur J Hum Genet 2010 Sep;18(9):Epub 2010 Apr 7 doi: 10.1038/ejhg.2010.42. PMID: 20372188 **Free PMC Article**
[Guidelines for the diagnosis and management of Marfan syndrome.](#)
Ades L; CSANZ Cardiovascular Genetics Working Group



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National Center for Biotechnology Information

NCBI Webinars

PubMed Clinical Queries about diagnosis, etiology, therapy, prognosis, systematic reviews

Recent clinical studies
Go to:

Etiology
[The effects of acute and elective cardiac surgery on the anxiety traits of patients with Marfan syndrome.](#)
Benke K, Ágg B, Pólos M, Sayour AA, Radovits T, Bartha E, Nagy P, Rákóczi B, Koller Á, Szokolai V, Hedberg J, Merkely B, Nagy ZB, Szabolcs Z
BMC Psychiatry 2017 Jul 17;17(1):253. doi: 10.1186/s12888-017-1417-9. PMID: 28716062 **Free PMC Article**
[Relationship between fibrillin-1 genotype and severity of cardiovascular involvement in Marfan syndrome.](#)
Franken R, Teikido-Tura G, Brion M, Forteza A, Rodriguez-Palomares J, Gutierrez L, Garcia Dorado D, Pals G, Mulder BJ, Evangelista A
Heart 2017 Nov;103(22):1795-1799. Epub 2017 May 3 doi: 10.1136/heartjnl-2016-310631. PMID: 28468757
[Pregnancy-related acute aortic dissection in Marfan syndrome: A review of the literature.](#)
Smith K, Gros B
Congenit Heart Dis 2017 May;12(3):251-260. Epub 2017 Apr 2 doi: 10.1111/chd.12465. PMID: 28371362
[Aortic events in a nationwide Marfan syndrome cohort.](#)
Groth KA, Stochholm K, Hove H, Kyhl K, Gregersen PA, Vejstrup N, Østergaard JR, Gravholt CH, Andersen NH
Clin Res Cardiol 2017 Feb;106(2):105-112. Epub 2016 Aug 22 doi: 10.1007/s00392-016-1028-3. PMID: 27550511
[Long-term outcomes of aortic root operations for Marfan syndrome: A comparison of Bentall versus aortic valve-sparing procedures.](#)
Price J, Magruder JT, Young A, Grimm JC, Patel ND, Alejo D, Dietz HC, Vricella LA, Cameron DE
J Thorac Cardiovasc Surg 2016 Feb;151(2):330-6. Epub 2015 Oct 27 doi: 10.1016/j.jtcvs.2015.10.068. PMID: 26704057
See all (1440)

Diagnosis
[The effects of acute and elective cardiac surgery on the anxiety traits of patients with Marfan syndrome.](#)
Benke K, Ágg B, Pólos M, Sayour AA, Radovits T, Bartha E, Nagy P, Rákóczi B, Koller Á, Szokolai V, Hedberg J, Merkely B, Nagy ZB, Szabolcs Z
BMC Psychiatry 2017 Jul 17;17(1):253. doi: 10.1186/s12888-017-1417-9. PMID: 28716062 **Free PMC Article**
[A Novel Fibrillin-1 Gene Mutation Leading to Marfan Syndrome in a Korean Girl.](#)
Nam HK, Nam MH, Ha KS, Rhie YJ, Lee KH
Ann Clin Lab Sci 2017 Mar;47(2):221-225. PMID: 28442527



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Genetic Testing Registry

Deletion/duplication analysis (85)

Detection of homozygosity (1)

Mutation scanning of the entire coding region (6)

Sequence analysis of select exons (13)

Sequence analysis of the entire coding region (134)

Targeted variant analysis (23)

See all (170)

Clinical resources

OMIM

Orphanet

ClinicalTrials.gov

Molecular resources

OMIM

View FBN1 variations in ClinVar

RefSeqGene

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Consumer resources

Genetic Alliance

Genetics Home Reference

MalaCards

MedlinePlus

NCATS Office of Rare Diseases Research (GARD)

PubMed Health

Marfan Syndrome

Medical Encyclopedia

Diseases & Conditions:

Mitral Valve Prolapse

Marfan Syndrome

Heart Valve Disease

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Reviews

GeneReviews

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Reviews in PubMed

Related information

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Functional Class

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GTR

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MeSH

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PubMed (Bookshelf cited)

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MedGen as terminology service

Aggregates and harmonizes genetic phenotype information

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- Phenotype backbone of
 - NIH Genetic Testing Registry (GTR, <https://www.ncbi.nlm.nih.gov/gtr>) test descriptions
 - ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar>) variant interpretations
- Computational access through ftp reports (<ftp://ftp.ncbi.nlm.nih.gov/pub/medgen/>) and NCBI's API, e-utilities (esearch and esummary)
- Hospital/clinic systems can use NCBI's API to tailor views of genetic information for their users at point of care

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How can MedGen help the clinical process?

- Prepare for a clinic visit
 - Research a phenotype: one page has all freely available information about a condition, phenotype or drug response from authoritative sources
 - Help suggest conditions that fit the cluster of clinical features for a patient
- During a clinic visit
 - List clinical features to evaluate a patient for a suspected diagnosis
 - Actionable links to professional practice guidelines, clinicaltrials.gov
 - Links to available tests for the condition
 - Therapeutic recommendations for drug responses based on genotype from professional societies
- After a clinic visit
 - Links to ClinVar for help with variant interpretation
- Links to consumer resources to educate the patient and family about the diagnosis

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- URLs & Places to Learn More -

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Questions?



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